

Claims

What is claimed is:

1. A method for interrogating genetic variations comprising:
5 obtaining a plurality of functional regions of the genome, wherein the functional regions comprise at least 10,000 bases;
determining sequence variations of a plurality of individuals in the functional regions of the genome.
- 10 2. The method of Claim 1 wherein the functional regions comprise a plurality of transcription factor binding sites.
3. The method of Claim 2 wherein the functional regions comprise a plurality of RNA:protein binding domains.
- 15 4. The method of Claim 3 wherein the functional regions comprise a plurality of chromatin modification sites.
5. The method of Claim 4 wherein the functional regions comprise a plurality of origins
20 of replication.
6. The method of Claim 5 wherein the functional regions comprise a plurality of DNA methylation sites.

7. The method of Claim 1 wherein the obtaining comprises determining functional regions using microarrays.
- 5 8. The method of Claim 7 wherein the microarrays are high density oligonucleotide arrays.
9. The method of Claim 8 wherein the microarrays comprise oligonucleotide probes tiling regions of the genome.
- 10 10. The method of Claim 9 wherein the determining comprises determining the sequences of the functional regions of a plurality of individuals.
11. The method of Claim 9 wherein the determining comprises determining the
15 genotypes of the functional regions of a plurality of individuals.
12. The method of Claim 11 wherein the genotypes are SNP genotypes.
13. The method of Claim 12 wherein the determining comprises performing a WGS
20 with at least one restriction enzyme that is suitable for interrogating at least one functional region.

14. The method of Claim 9 wherein the determining comprises determining sequence copy number changes.
15. The method of Claim 1 wherein the functional regions comprise at least 100000 bases.
16. The method of Claim 15 wherein the functional regions comprise at least 500000 bases.
17. A method for interrogating genetic variations comprising:
obtaining at least one interested genomic segment;
obtaining a plurality of functional regions within the interested genomic segment,
wherein the functional regions comprise at least 5,000 bases;
determining sequence variations of a plurality of individuals in the functional regions of the genome.
18. The method of Claim 17 wherein the interested genomic region is determined by association or linkage analysis.
19. The method of Claim 18 wherein the functional regions comprise a plurality of transcription factor binding sites.

20. The method of Claim 18 wherein the functional regions comprise a plurality of RNA:protein binding domains.
21. The method of Claim 18 wherein the functional regions comprise a plurality of chromatin modification sites.
22. The method of Claim 18 wherein the functional regions comprise a plurality of origins of replication.
23. The method of Claim 18 wherein the functional regions comprise a plurality of DNA methylation sites.
24. The method of Claim 17 wherein the obtaining comprises determining functional regions using microarrays.
25. The method of Claim 24 wherein the microarrays are high density oligonucleotide arrays.
26. The method of Claim 25 wherein the microarrays comprise oligonucleotide probes tiling regions of the genome.
27. The method of Claim 24 wherein the determining comprises determining the sequences of the functional regions of a plurality of individuals.

28. The method of Claim 24 wherein the determining comprises determining the genotypes of the functional regions of a plurality of individuals.

5 29. The method of Claim 28 wherein the genotypes are SNP genotypes.

30. The method of Claim 29 wherein the determining comprises performing a WGS with at least one restriction enzyme that is suitable for interrogating at least one functional region.

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31. The method of Claim 24 wherein the determining comprises determining sequence copy number changes.